
Noonan syndrome

Summary

- Noonan syndrome is a genetic condition that typically includes heart abnormalities and characteristic facial features.
 - About one-third of affected children have mild intellectual disability.
 - Noonan syndrome may be inherited in up to 75 per cent of cases.
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Noonan syndrome is a genetic condition that usually includes heart anomalies and characteristic facial features. About one third of affected children have mild intellectual disability. Noonan syndrome varies from relatively mild to severe. The physical characteristics are not always obvious and can be hard to identify in individuals with more subtle traits.

Estimates vary, but it is thought that between one in 1,000 to one in 5,000 children have Noonan syndrome, with males and females equally affected. Genetic changes on several genes are known to cause Noonan syndrome, but the diagnosis is usually established on the basis of clinical features.

Currently there is no cure, but symptoms of Noonan syndrome can be medically managed. Your child's health care team will coordinate various management strategies for different symptoms. These can include cardiac monitoring for heart irregularities, medication for blood clotting, and speech and educational interventions for any learning difficulties they may experience.

Effects of Noonan syndrome

Parts of the body typically affected by Noonan syndrome include:

- heart
- facial features
- musculoskeletal system
- kidneys, spleen, genitals and liver.

Associated heart conditions

About 65 per cent of children with Noonan syndrome have heart anomalies. The most common include:

- **pulmonary valve stenosis** – the pulmonary valve allows blood with reduced oxygen content (deoxygenated blood) to leave the heart and go to the lungs. Pulmonary valve stenosis occurs when the pulmonary valve is narrower than usual
- **hypertrophic cardiomyopathy** – thickening of the heart muscles
- **atrial septal defect** – a hole between the two upper chambers of the heart (atria)
- **ventricular septal defect** – a hole between the two lower chambers of the heart (ventricles).

Characteristic facial features

Some facial features are characteristic of Noonan syndrome. How these features are expressed in children with Noonan syndrome varies from one child to the next and can change with age. Characteristics may include:

- heavy or hooded eyelids that may interfere with vision (ptosis)
- downward sloping eyes with an extra fold of skin at each inner corner
- widely spaced eyes (hypertelorism) with a flattened bridge of the nose
- brilliant blue or blue-green eyes

- strongly arched (diamond-shaped) eyebrows
- low-set ears, tilted back so that the lobes point forward more than usual
- low hairline at the neck
- coarse and curly hair
- short neck with extra skin folds or 'webbing' at the back of the neck – this makes the neck look wider when viewed from behind.

Associated musculoskeletal conditions

Some possible anomalies of the musculoskeletal system include:

- short stature – although birth weight is usually average, children with Noonan syndrome are frequently shorter than expected for their age. As adults, people with Noonan syndrome are often short – men average 165 cm (5' 5") and women 152 cm (5') in height
- low muscle tone and impaired coordination
- unusually shaped breastbone (sternum) – may be sunken or raised
- curvature of the spine (scoliosis).

Associated kidney, spleen, genital and liver conditions

Noonan syndrome can also affect the kidneys, spleen, genitalia and liver:

- Puberty can be delayed in both males and females.
- Females are normally fertile, however up to 80 per cent of males can have reduced fertility due to deficient sperm production, undescended testicles or testicular dysfunction.
- Approximately 25 to 50 per cent of people with Noonan syndrome can develop an enlarged liver or spleen.
- Less than 10 per cent of people with Noonan syndrome will develop mild kidney problems.

Learning and language issues with Noonan syndrome

Up to 35 per cent of people with Noonan syndrome will have a mild intellectual disability, but most people will be unaffected.

People who experience intellectual disability with Noonan syndrome may also experience developmental delays with their speech, language and coordination. They may also exhibit some mild emotional and behavioural issues.

Other issues associated with Noonan syndrome

Depending on the severity, other issues experienced by people with Noonan syndrome may include:

- eye problems, including squint and short-sightedness
- babies may have feeding problems and may vomit easily
- hearing problems, including a susceptibility to glue ear
- delay in reaching developmental milestones, such as sitting unaided and walking
- easily bruised skin and susceptibility to nose bleeds
- reduced resistance to infection
- dental problems, including missing teeth
- increased risk of seizures
- undescended testicles in males
- decreased fertility in males who have had undescended testicles
- delayed puberty.

Cause of Noonan syndrome

Noonan syndrome is a genetic condition caused by a change in one of at least seven different genes. Genetic testing has shown that a change in the *PTPN11* gene causes Noonan syndrome in about 50 per cent of affected people.

It was once believed that most cases of Noonan syndrome were sporadic, which means the child's gene spontaneously changed. However, researchers now suspect that Noonan syndrome is inherited in up to 75 per cent of cases.

The pattern of inheritance is autosomal dominant, which means that the gene (and the condition) can be transmitted from parent to child. Each child of a parent who carries the gene has a 50 per cent chance of inheriting the gene and developing the condition.

Diagnosis of Noonan syndrome

Diagnosis is generally made by noting the presence of typical Noonan syndrome features and ruling out other possible causes, such as **fetal alcohol spectrum disorder**, **neurofibromatosis type 1** (also known as Von Recklinghausen's disease, or NF1) and **Turner's syndrome**.

Genetic testing is available for Noonan syndrome and options can be discussed with your health professional.

Treatment for Noonan syndrome

There is currently no cure for Noonan syndrome. Treatment and condition management aims to ease some of the associated issues. If your child has been diagnosed with Noonan syndrome their treatment may include:

- for a mild heart irregularity, it may be enough to carefully monitor your child on an outpatient basis
- surgical correction of any severe heart structural anomalies
- if feeding is a problem, your baby may be fed by nasogastric tube (a tube threaded into the nose, down the oesophagus and into the stomach)
- prescription glasses – to correct short-sightedness
- regular injections of growth hormone – to help your child achieve a height closer to that typical for their age
- surgical correction of undescended testicles (orchidopexy)
- anticonvulsant medication – if your child experiences seizures
- speech therapy
- special education – this is necessary for about 10 per cent of children with Noonan syndrome
- behaviour management – if appropriate
- treatment to manage mild blood-clotting problems
- dental and orthodontic treatment
- if glue ear is a problem, it can be managed with an operation to insert drainage tubes (grommets).

Genetic counselling and Noonan syndrome

If you, your child or another family member have been diagnosed with Noonan syndrome, or if Noonan syndrome runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand a condition and what causes it, how it is inherited, and what a diagnosis means for your or your child's health and development for the future. Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

If a Noonan syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

The **Noonan Syndrome Awareness Association** website provides information about the condition as well as Noonan syndrome research, resources and support.

Where to get help

- Your **GP (doctor)**
- **Victorian Clinical Genetics Services (VCGS)**, Royal Children's Hospital Tel. **1300 118 247**

betterhealth.vic.gov.au

- **Genetic Support Network of Victoria** Tel. **(03) 8341 6315**
- **Noonan Syndrome Awareness Association**

This page has been produced in consultation with and approved by:

Victorian Clinical Genetics Services (VCGS)

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